hypochondroplasia

Hypochondroplasia is a form of short-limbed dwarfism. This condition affects the conversion of cartilage into bone (a process called ossification), particularly in the long bones of the arms and legs. Hypochondroplasia is similar to another skeletal disorder called achondroplasia, but the features tend to be milder.

All people with hypochondroplasia have short stature. The adult height for men with this condition ranges from 138 centimeters to 165 centimeters (4 feet, 6 inches to 5 feet, 5 inches). The height range for adult women is 128 centimeters to 151 centimeters (4 feet, 2 inches to 4 feet, 11 inches).

People with hypochondroplasia have short arms and legs and broad, short hands and feet. Other characteristic features include a large head, limited range of motion at the elbows, a sway of the lower back (lordosis), and bowed legs. These signs are generally less pronounced than those seen with achondroplasia and may not be noticeable until early or middle childhood. Some studies have reported that a small percentage of people with hypochondroplasia have mild to moderate intellectual disability or learning problems, but other studies have produced conflicting results.

Frequency

The incidence of hypochondroplasia is unknown. Researchers believe that it may be about as common as achondroplasia, which occurs in 1 in 15,000 to 40,000 newborns. More than 200 people worldwide have been diagnosed with hypochondroplasia.

Genetic Changes

About 70 percent of all cases of hypochondroplasia are caused by mutations in the *FGFR3* gene. This gene provides instructions for making a protein that is involved in the development and maintenance of bone and brain tissue. Although it remains unclear how *FGFR3* mutations lead to the features of hypochondroplasia, researchers believe that these genetic changes cause the protein to be overly active. The overactive FGFR3 protein likely interferes with skeletal development and leads to the disturbances in bone growth that are characteristic of this disorder.

In the absence of a mutation in the *FGFR3* gene, the cause of hypochondroplasia is unknown. Researchers suspect that mutations in other genes are involved, although these genes have not been identified.

Inheritance Pattern

Hypochondroplasia is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Most people with hypochondroplasia have average-size parents; these cases result from a new mutation in the *FGFR3* gene. In the remaining cases, people with hypochondroplasia have inherited an altered *FGFR3* gene from one or two affected parents. Individuals who inherit two altered copies of this gene typically have more severe problems with bone growth than those who inherit a single *FGFR3* mutation.

Other Names for This Condition

- HCH
- Hypochondrodysplasia

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Hypochondroplasia
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0410529/

Other Diagnosis and Management Resources

- GeneReview: Hypochondroplasia
  https://www.ncbi.nlm.nih.gov/books/NBK1477
- MedlinePlus Encyclopedia: Lordosis
  https://medlineplus.gov/ency/article/003278.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Lordosis
  https://medlineplus.gov/ency/article/003278.htm
- Health Topic: Dwarfism
  https://medlineplus.gov/dwarfism.html

Genetic and Rare Diseases Information Center

- Hypochondroplasia
  https://rarediseases.info.nih.gov/diseases/6724/hypochondroplasia

Educational Resources

- Disease InfoSearch: Hypochondroplasia
  http://www.diseaseinfosearch.org/Hypochondroplasia/3635
- KidsHealth from the Nemours Foundation
  http://kidshealth.org/en/parents/dwarfism.html
- MalaCards: hypochondroplasia
  http://www.malacards.org/card/hypochondroplasia
- My46 Trait Profile
  https://www.my46.org/trait-document?trait=Hypochondroplasia&type=profile
- Orphanet: Hypochondroplasia
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=429

Patient Support and Advocacy Resources

- Human Growth Foundation
  http://hgfound.org/
- International Skeletal Dysplasia Registry, UCLA
  http://ortho.ucla.edu/isdr
- Little People of America, Inc.
  http://www.lpaonline.org
- National Organization for Rare Disorders
  https://rarediseases.org/rare-diseases/hypochondroplasia/
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/dwarfism.html
- The MAGIC Foundation
  https://www.magicfoundation.org/

GeneReviews

- Hypochondroplasia
  https://www.ncbi.nlm.nih.gov/books/NBK1477

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22hypochondroplasia%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Dwarfism%5BMAJR%5D%29+AND+%28%28hypochondroplasia%5BTIAB%5D%29+OR+%28hch%5BTIAB%5D%29+OR+%28hypochondrodysplasia%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- HYPOCHONDROPLASIA
  http://omim.org/entry/146000

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